MODERN METHODS OF NEONATAL SCREENING

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Neonatal screening is a program, which aims at early identification of conditions for which early and timely intervention can prevent or reduce associated mortality and morbidity. The most modern screening technology is the tandem mass spectrometry (tandem MS; MS/MS). MS/MS-newborn screening requires confirmatory testing and clinical evaluation before a diagnosis can be made.

Before starting the neonatal screening with this high technology system you have to make sure that long-term follow up monitoring and management is guaranteed by clinical professionals and respective facilities and resources for medical food, medications and supplements required for treatment are available.

Recommended neonatal screening done by “one test, one disorder tests” (like screening for hypothyroidism or biotinidase deficiency) as well as by MS/MS technic is different in the different countries in Europe, USA, Australia and Canada. The German recommendation for newborn screening in cludes (since 2004): Hypothyroidism; Congenital adrenal hyperplasia (CAH); Biotinidase Deficiency; Galactosaemia (Classic); Phenylketonuria (PKU); Maple syrup urine disease (MSUD); Medium Chain Acyl-CoA Dehydrogenase (MCAD)-Deficiency; Long-Chain-3-0H Acyl-CoA-Dehyd rogenase (LCHAD)-Deficiency; (Very-) Long-Chain Acyl-CoA-D ehydrogenase (VLCAD)-Deficiency; Carnitin-Palmitoyl-CoA-Transferase I (CPT I)-Deficiency; Carnitin-Palmitoyl-CoA-Transferase II (CPT II)-Deficiency; Carnitin-Acylcarnitin-Tran slocase (CACT)-Deficiency; Glutaric acidaemia type I (GA 1).

All inborn errors – except the 4 listed on top – can be detected by MS/MS-Screening. All of the nearly 700.00 newborn babies per year in Germany are checked under the same conditions and under strong administ rative and quality controls. The expansion of the neonatal screening panel by MS/MS and other methods is under discussion. Target disorders are: Cystic fibrosis (CF); Methyl malonic acidaemia (different types); Tyrosinosis type I; Argininosuccinic aciduria; Lysosomal storage Disorders (now treatable) like Morbus Pompe, Hunter- and Hurler Diseases, Morbus Gaucher and Fabry Disease. Details of the discussion about new screening recommendations were given. Molecular genetic tests are often used in Germany for the confirmation diagnostics. The Ukraine recommendation for newborn screening includes only Hypothyroidism, Congenital adrenal hyperplasia (CAH), Mucoviscidosis Phenylketonuria (PKU). So, maybe we need more for diagnosis of inherited pathology.

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